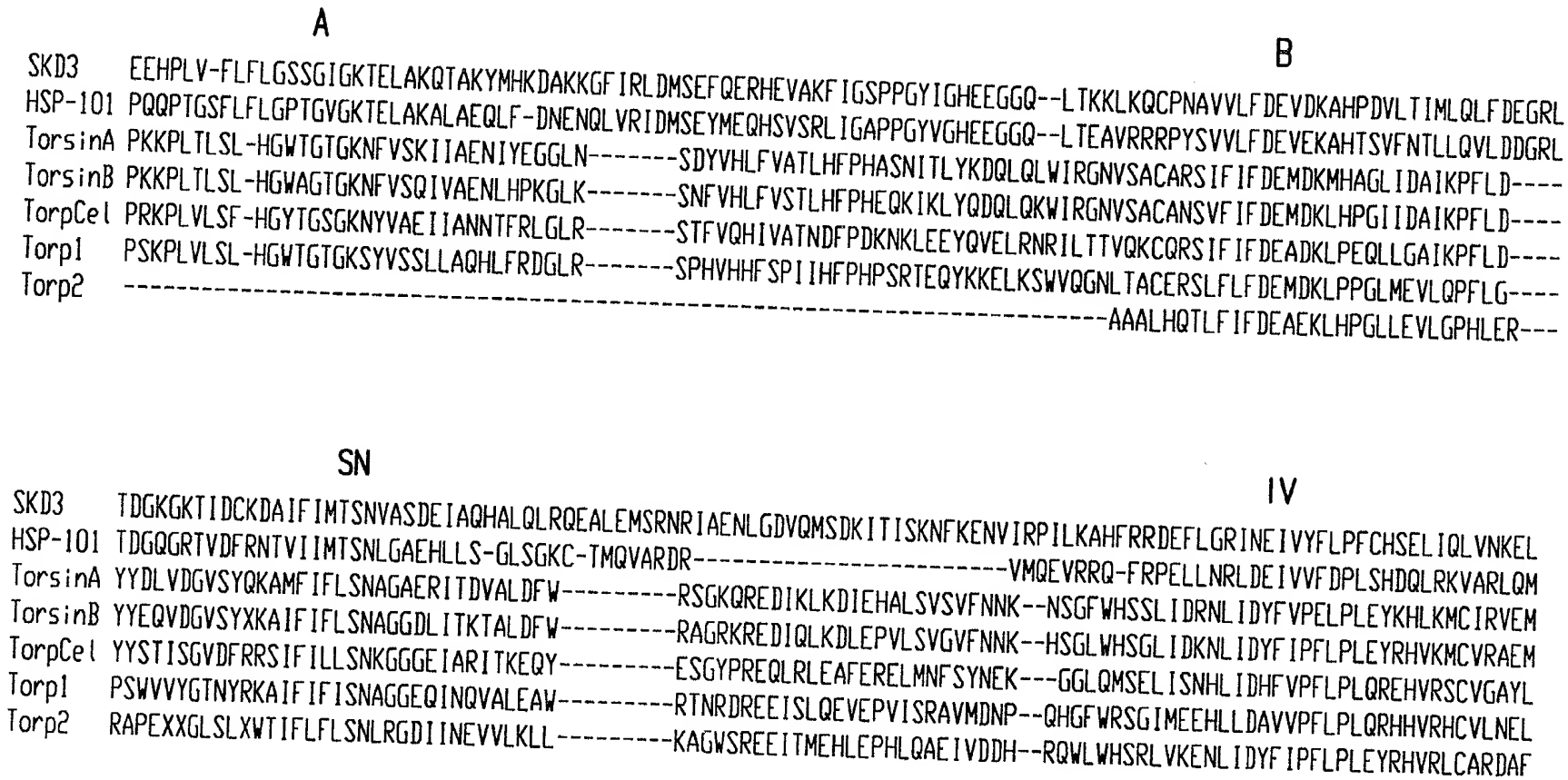


FIG. 4C

FIG. 5B

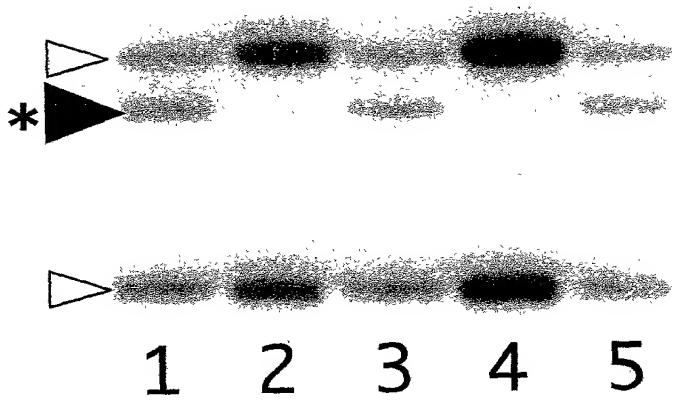


FIG. 5C

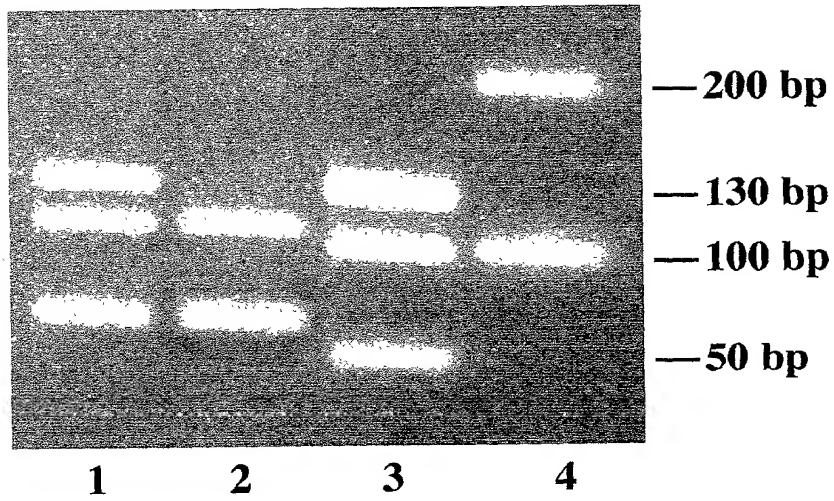
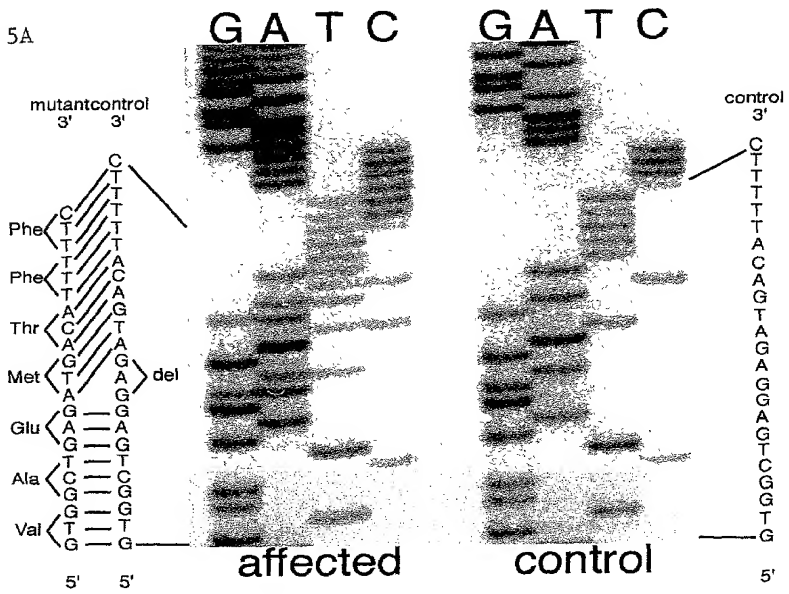


FIG. 5A



6419

CCTGGAATACAAACACCTAAAAATGTGTATCCGAGTGGAAATGCAGTCCCGAGGCTATGAAATTGATGAAGACATTGTAAGCAGA
GGACCTTATGTTTGTGGATTTTTACACATAGGCTCACCTTTACGTCAGGGCTCCGATACTTTAACTACTTCTGTAACATTCGTCT

BseRI* BseRI

GTGGCTGAGGAGATGACATTTTTCCCCAAAGAGGAGAGAGTTTTCTCAGATAAAGGCTGCAAAACGGTGTTCACCAAGTTAGATT
CACCGACTCCTCTACTGTAAAAAGGGGTTTCTCCTCTCTCAAAAGAGTCTATTTCCGACGTTTTGCCACAAGTGGTTCAATCTAA

ATTACTACGATGATTGACAGTCATGATTGGCAGCCGGAGTCACTGCCTGGAGTTGGAAAGAAACAACACTCAGTCCTTCCACC
TAATGATGCTACTAACTGTCAGTACTAACCGTCGGCCTCAGTGACGGACCTCAACCTTTCTTTGTTGTGAGTCAGGAAGGTGG

H48 6418

FIG. 5D

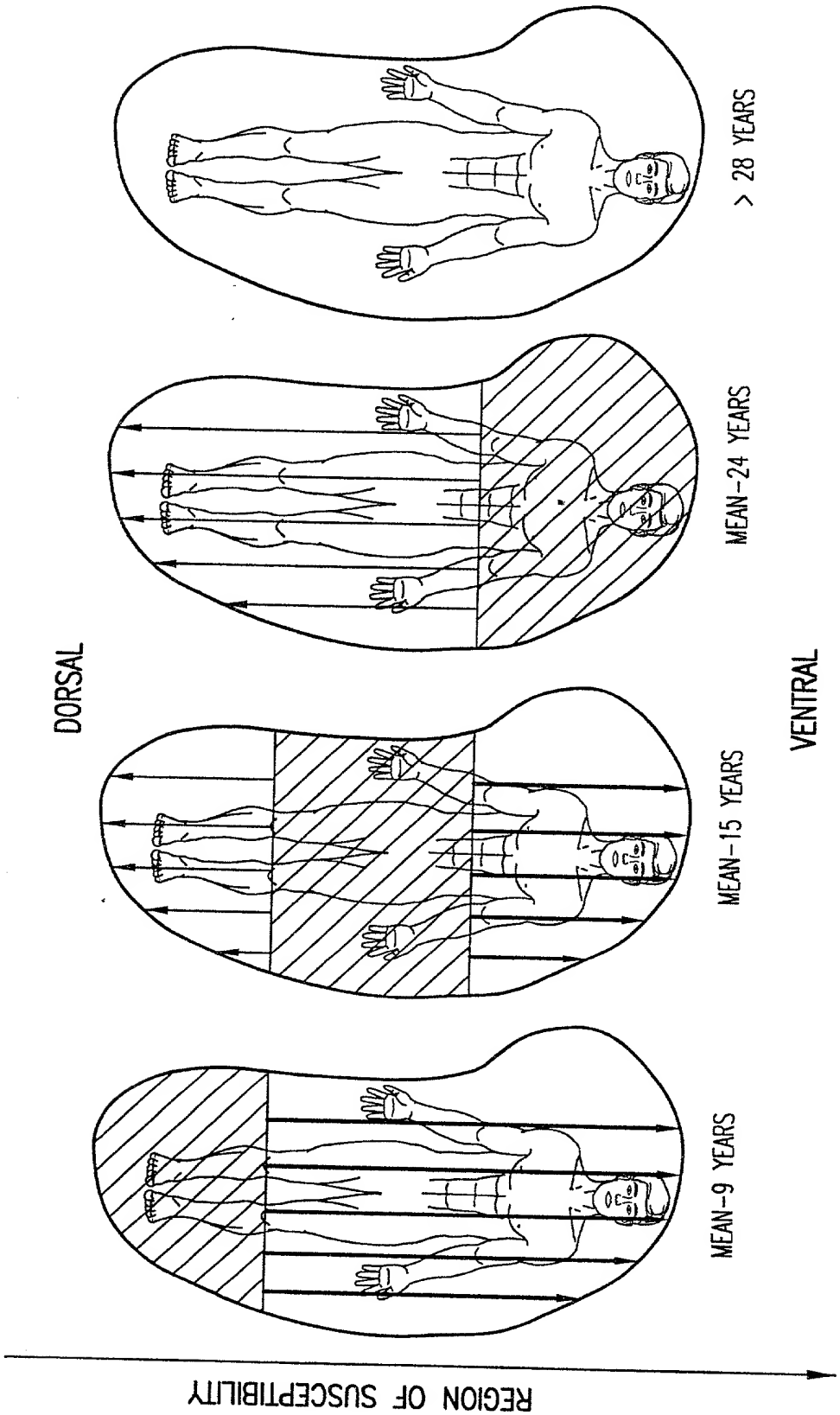

DORSAL
VENTRAL
MEAN-9 YEARS
MEAN-15 YEARS
MEAN-24 YEARS
> 28 YEARS
AGE
REGION OF SUSCEPTIBILITY

FIG.6

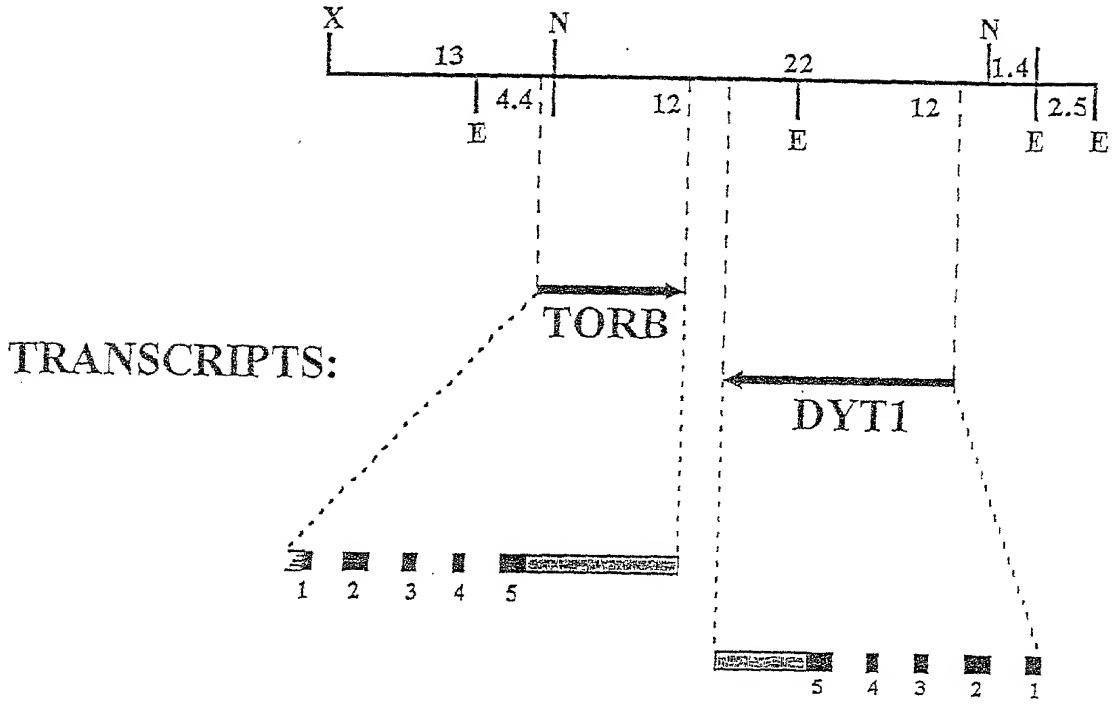

X
N
N
13
22
1.4
4.4
12
12
2.5
E
E
E
E
TRANSCRIPTS:
TORB
DYT1
1 2 3 4 5
5 4 3 2 1

FIG. 7

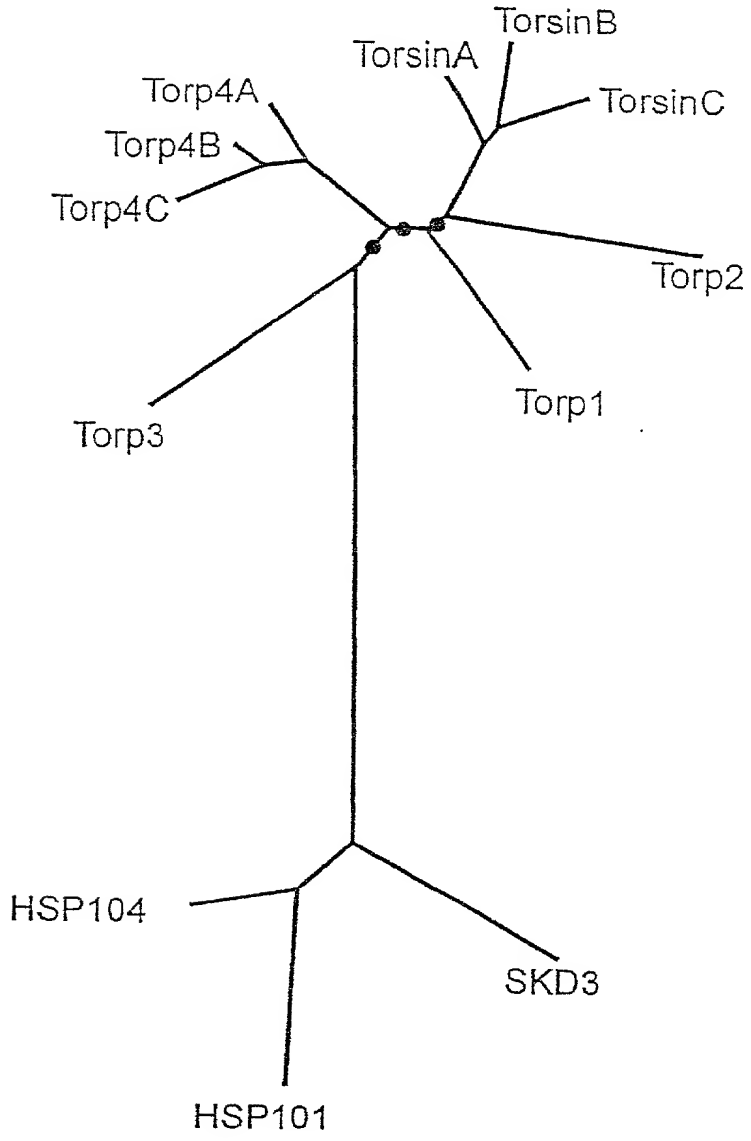

TorsinB
TorsinA
Torp4A
TorsinC
Torp4B
Torp4C
Torp2
Torp1
Torp3
HSP104
SKD3
HSP101

FIG. 8

FIG. 9A

DYTI

Exon	Size (bp)	3'end of exon	5' splice site (SEQ ID NO)	Intron (kb)[a]	3' splice site (SEQ ID NO)	5' end of exon (SEQ ID NO)	Exon
1	178	...CGG GAG G R E A	GTAGGCTGGG... (67)	1.2	...TCTTTCCCAG (71)	CA CTG CAG AAG... (75) L G K	2
2	266	...TTG TAC AAG L Y K	GCAAGGATGG...(68)	1.5	...TTTAATTCAG (72)	GAT CAG TTA... D Q L	3
3	176	...TTT CTC AG F L S	GTAAGGTCAG... (69)	0.097	...TGTTTTGCAG (73)	C AAT GCT GGA... (76) N A G	4
4	128	...AAG AAC AG K N S	GTGAGTAGGG... (70)	4	...TTCTTCCCAG (74)	T GGC TTC TGG... (77) G F W	5
5	251	...GAT GAT TGA D D *					

FIG. 9B

TORB

Exon	Size (bp)	3'end of exon	5' splice site (SEQ ID NO)	Intron (kb)[a]	3' splice site (SEQ ID NO)	5' end of exon (SEQ ID NO)	Exon
1	n.d.	...GCT TCG G A S A		n.d.	...GTTCTTGCAG (81)	CT CTC AAG CTG... (85) L K L	2
2	266	...CTG TAC CAG L Y Q	GCAAGAGAAC... (78)	3	...GTTGGTCCAG (82)	GAC CAG TTA... D Q L	3
3	176	...TTT CTC AG F L S	GTCAGCGGGA... (79)	1.8	...GCAAACTCAG (83)	C AAT GCA GGC... (86) N A G	4
4	128	...AAA CAC AG K H S	GTGAGTCCAC... (80)	.31	...TGTTCTGCAG (84)	T GGC CTG TGG... (87) G L W	5
5	242	...TTC CAC TGA F H *					

*Sizes of introns were approximated by gel resolution of PCR products

FIG. 10

Type	*Number of Individuals*	*Age of onset yrs*	*Site of onset*[a] *(no.)*	*Sites involved (no.)*
Early onset:				
AJ	5	6, 8, 8, 10, 19	A(4), ANL(1)	A(7), N(3), T(1), L(2)
NJ	12	3, 6(2), 8, 9, 10, 11, 13, 14(2), 16, 18	A(9), AU(1)	A(20), G(9),
			G(1), L(1)	R(6), K(4), U(4), L(4),
				N(6), P(1), S(1), T(2),
				F(2), J(3), U(2)
Potential homozygosity:				
AJ[b]	5	4, 6, 7, 26, 35	L(4), M(1)	L(4), M(1), N(2), P(1),
				T(1), U(1), H(2)
Late onset:				
AJ	1	66	U	U

[a] Body sites: U=upper face, F=lower face, J=jaw, T=tongue, P=pharynx, L=larynx, S=speech, swallowing, A=arm, K=trunk, G=leg, N=neck, H=hearing loss. (no.=Number of individuals in group with that site affected.)

[b] Three of these individuals had all known AJ ancestors, one was 7/8 AJ, and one was 1/2 AJ.

	Genotype			
Sample number	*D9S2160*[a]	*D9S2161*	*D9S63*	*D9S2162*
18843	3,4	2,2	8,8	4,4
13709	4,4	2,6	14,14	NT[b]
14122[c]	3,3	4,4	0,0	5,5
13945	4,4	1,5	18,18	NT[b]
14040	2,5	1,2	16,16	2,4

[a] Markers listed from centromere to telomere: D9S2160 - <40 kb - D9S2161 - 150 kb - D9S63 - 130 kb - D9S2162 (Ozelius et al., 1997a).
* Position of DYT1 gene
[b] Not tested.
[c] This individual had onset at 7 years in the arm with eventual involvement of the arm and neck and a positive family history of movement disorder (father with tremor). He was also apparently homozygous for markers D9S159-D9S2158-D9S2159, which are proximal to DS2160 (total region of 100 kb).

FIG. 11

FIG. 12A

Intron Primers Used to Amplify DYT1 Exons			
Exon	Primer Sequence (5' → 3')	SEQ. ID NO.:	Product Size (bp)*
1	GCAAAACAGGGCTTTGTACCG	(SEQ. ID NO.: 30)	
	AGTAGAGACGCGGGTAGATG	(SEQ. ID NO.: 31)	
	GCGTCTCTACTGCCTCTTCG	(SEQ. ID NO.: 32)	
	ATGCCCTGGTCCTAGTTCAG	(SEQ. ID NO.: 33)	
2	GGTTTCGCAAGGTGCTTGGA	(SEQ. ID NO.: 34)	408
	GGGATTCCAAACTTCCATCC	(SEQ. ID NO.: 35)	
3 and 4	TCCATGGGGTTGGTAGGAAC	(SEQ. ID NO.: 36)	804
	GGTGACAGAGTAAAACTATCTG	(SEQ. ID NO.: 37)	
5	GACCCCCAGTAGACGTTTGT	(SEQ. ID NO.: 38)	640
	GTAAAAAATCATGAGCCCTGC	(SEQ. ID NO.: 39)	

FIG. 12B

Intron Primers Used to Amplify TORB Exons			
Exon	Primer Sequence (5' → 3')	SEQ. ID NO.:	Product Size (bp)*
1	n.d.#		-
2	CCAGAGTTAGTGAGCAGGTC	(SEQ. ID NO.: 40)	526
	GAAGCGTTAAGGACCTCCAC	(SEQ. ID NO.: 41)	
3	ATCTATCTCTGCCAATTTCCAC	(SEQ. ID NO.: 42)	466
	GTCCTGGTAAACAAAGTGCTG	(SEQ. ID NO.: 43)	
4	TGGGGTTACTCTATGTTGGTC	(SEQ. ID NO.: 44)	440
	CTAGCACAGTATGCCCTAAG	(SEQ. ID NO.: 45)	
5	TGAGGAATGTGCTGAGGGTC	(SEQ. ID NO.: 46)	333
	GCTGTCTCCTACCCCATCTG	(SEQ. ID NO.: 47)	

*PCR products were generated using oligonucleotides synthesized from intronic sequences, and accordingly the size of each product includes both intron and exon sequence.

#Not done. It was not possible to identify primers which could consistently PCR this exon.

Protein	*Organism*	*Gene*	*Locus*	*Accession*	*UniGene*
hTorsinA	human	DYT1	Chr.9, D9S159-D9S164	AF007871	Hs.19261
mTorsinA	mouse			AA230756	Mm.40438
rTorsinA	rat			AA850233	Rn.20041
sTorsinA	pig			AU058534	
hTorsinB	human	TORB	Chr.9, D9S159-D9S164	AF007872	Hs.5091
mTorsinB	mouse			AA596988	
drTorsinC	zebrafish			AA542632	
hTorp1	human	TORP1		AA873275	Hs.59038
mTorp1	mouse			AA981789	Mm.33875
rTorp1	rat			H31561	
hTorp2	human	TORP2		AA150869	Hs.26267
mTorp2	mouse			AA791729	
dmTorp3	fruitfly	EG:84H4.1	DMC84H4	AL031766	
ceTorp4A	nematode	F44G4.1	CEC18E9	P54073	
ceTorp4B	nematode	Y37A1B.12	CEY37A1B	AL023835	
ceTorp4C	nematode	Y37A1B.13	CEY37A1B	AL023835	

FIG. 13

INTRON 1 OF DYT1 GENE

FIG. 14A: Clone 1:
23g14-2-7050.cDNA (Length: 283) SEQ ID NO.: 48

```
1    gtaggctggg gcgggggctg gaggctgggg ctggggctgg ggctgggcga
51   tggcactagg gctgaactag gaccagggca tggagaatgg aggatggagg
101  ccgggggatg gcaccagggc cgggctagga ctagggctgg agcgggcct
151  gggggctggg gctgggcgat ggcactaggg cgggttgggg ctggggctgg
201  ggctgggggа tggagcgggg ccgggggctg ggggtggggc tgggggatcg
251  actagggctg gnttaggacc aggcggttgg cat
```

Bold = primer 4 (reverse sequence) from FIG. 12A
Underline = 5' splice sequence from Intron 1 FIG. 9A

FIG. 14B: Clone 2:
Harvey7-23g14-2.cDNA (Length: 375) SEQ ID NO.: 49

```
1    ggatggtgga tggaggctgg gggatggcag tagggccggg ctaggactgg
51   ggctggagcg gagtttgggg ctggggctca ggagcggggg ctggggctgg
101  ggctggggct gggggatggc actagggcag gccggggtag gggtcacatc
151  ccaggagggc cgggctgggc agagctgagt ccgcgggggc cggaccccgg
201  aagccaagcn gccggcctgc aggatgaggc ctggctcctc ggccatgacc
251  acagacgtgc cagacttaag tacggagacc tgaggagcca ggctgcagtt
301  ggcctacttt ncnctaagct gggggtggac cagtggtaac ctcctccgaa
351  gtgggttctg ctctttctag cctag
```

FIG. 14C: Clone 3:
23g1-Harvey11.cDNA (Length: 439) SEQ ID NO.: 50

```
1    ccactgccac tgccaccagt ttgcacccct aacccctgtn ctgctcctcc
51   caccccaagg cagagccggn gaaaggaaac agtttggtcc ctcctggtcg
101  gctgcggaag agtctcacca tccttctgtc tccgtagcta gaaaggaggc
151  agaacccaca ttcggaggga ggttaccact ggtccacccc cagcttagcg
201  caaagtaggc caacctgcat gcctggnnct cctcaggntc tgcctactta
251  agtctggcag ctctnnnntca tggccgaggt agccaggctc atcctgcagg
301  nnccngccnn ttgncttncc ggggtntcgn nnccccgtac tcagctcgtc
351  cagccggcct ctggatgtga cctaccgctg ctagtgcatc ccagccagcc
401  agccagccgt ctagccagcc aactgctcag ccagtctag
```

FIG. 14D: Clone 4:
23g1-Harvey6.cDNA (Length: 378) SEQ ID NO.: 51

```
1    ctgggaaaga caaagccaat caggagtggg gaagaaacac ggcaaaatgt
51   agccacattt acagcccata aganagccag caaagccgtc tagcctccaa
101  gcaccttgcg aaacctcaag tactgcggtc tggtaagctc ctggcccaga
151  ggggacggcg gtccagggng ccctcccttt gctggtcctg cctattctaa
201  agccctggcc cgnctccttc ccgaaaagcc ccttggtgcc actgccactg
251  ccaccanttt gcncccctac ccctgtnctg ctcctcccac cccaaggcag
301  atgcggnngg ngaaaggaaa cantttggtc cctcctggtc ggctcgngga
351  agactcctca ccatccttcc tgtcttcc
```

Bold = primer 5 from FIG. 12A
Italics = sequence overlap between Clone 4 and Clone 3
Underline = 3' splice site from FIG. 9A

FIG. 14E: Clone 4:
23g1-Harvey6.cDNA (Length: 388) SEQ ID NO.: 88

1 ctgggaaaga ctgggaaaga caaagccaat caggagtggg gaagaaacac
51 ggcaaaatgt agccacattt acagcccata aganagccag caaagccgtc
101 tagcc**tccaa gcaccttgcg aaacc**tcaag tactgcggtc tggtaagctc
151 ctggcccaga ggggacggcg gtccagggng ccctcccttt gctggtcctg
201 cctattctaa agccctggcc cgnctccttc ccgaaaagcc ccttggtgcc
251 *actgccactg ccaccanttt gcnccccctac ccctgtnctg ctcctcccac*
301 *cccaaggcag atgcggnngg ngaaaggaaa cantttggtc cctcctggtc*
351 *ggctcgngga agactcctca ccatccttcc tgtcttcc*

Bold = primer 5 from FIG. 12A
Italics = sequence overlap between Clone 4 and Clone 3
Underline = 3' splice site from FIG. 9A

INTRON 2 DYT1 GENE

FIG. 15A: Clone 1:
29a5-6343.cDNA (Length: 400) SEQ ID NO.: 52

```
1    gaatatttac gagggtggtc tgaacagtga ctatgtccac ctgtttgtgg
51   ccacattgct ctttccacat gcttcaaaca tcaccttgta caaggcaagg
101  atggaagttt ggaatcccct cctggatgtc atcgggtttg gggtctcttt
151  gttgtgggat gagatttggg agttctatgt tgaaatgagt gagcccggaa
201  aacggttcat gtctcagttc cccttggaaa ggtgtagaag ttaagagttt
251  gagatgcgtg gagcagttaa taccatcaaa gctttgtggt gggttctgaa
301  aatcggtcca gtgagtatgt agggtcatgg gattttagag gtggacatga
351  tcaaatccat cttagagatc aacacatctc actcattttt attttcttat
```

Bold = primer 6 from FIG. 12A
Underline = 5' splice site sequence for intron 2 from FIG. 9A

FIG. 15B: Clone 1:
29a5-6343.cDNA (Length: 402) SEQ ID NO.: 89

```
1    gaatatttac gagggtggtc tgaacagtga ctatgtccac ctgtttgtgg
51   ccacattgct ctttccacat gcttcaaaca tcaccttgta caaggcaagg
101  atggaagttt ggaatcccct cctggatgtc atcgggtttg gggtctcttt
151  gttgtgggat gagatttggg agttctatgt tgaaatgagt gagcccggaa
201  aacggttcat gtctcagttc cccttggaaa ggtgtagaag ttaagagttt
251  gagatgcgtg gagcagttaa taccatcaaa gctttgtggt gggttctgaa
301  aatcggtcca gtgagtatgt agggtcatgg gattttagag gtggacatga
351  tcaaatccat cttagagatc aacacatctc actcattttt attttcttat
401  tt
```

Bold = primer 6 from FIG. 12A
Underline = 5' splice site sequence for intron 2 from FIG. 9A

FIG. 15C: Clone 2:
6550-54a5s.cDNA (Length: 418) SEQ ID NO.: 53

```
1    tttggagtga gacaggactg ggttcaggtc ccagctctgc cacatatagt
51   cttgggcaag tggagtaagc gctctctgtg cctcagttcc ctcatctgta
101  aaatgagaac gatagtgccc actccatggg gttggtagga acaaagaaga
151  ttttgggcat gtaaagttct tagtgccgag tgcacagtgg tctgtaagtg
201  aagctgcggt tcttagtggt agaaggagct gattgatggc cctggctgag
251  aactttgtgt tcgctttttc ccnttttaat tcaggatcag ttacagttgt
301  ggattcgagg caacgtgagt gcctgtgcga ggtccatctt catatttgat
351  gaaatggata agatgcatgc aggcctcata gatgccntca ancctttcct
401  cgactattat gacctggt
```

Bold = primer 7 from FIG. 12a
Underline = 3' splice sequence for intron 2 from FIG. 9A
Italics = EXON 3 sequence

INTRON 3 DYT1

FIG. 16A: Clone 1:
6202-54a5.cDNA (Length: 198) SEQ ID NO.: 54

1 **ctcgactatt atgacctggt ggatggggtc tcctaccaga aagccatgtt**
51 **catatttctc** aggtaaggtc agggctagga catgatggat gggccccgag
101 cccaagcctc tgagctccag gagaaaaccc tgtccttacc cactgggatt
151 gttttgcagc **aatgctggag cagaaaggat cacagatgtg tttggatt**

FIG. 16B: Clone 1:
6202-54a5.cDNA (Length: 200) SEQ ID NO.: 90

1 **ctcgactatt atgacctggt ggatggggtc tcctaccaga aagccatgtt**
51 **catatttctc** aggtaaggtc agggctagga catgatggat gggccccgag
101 cccaagcctc tgagctccag gagaaaaccc tgtccttacc cactgggatt
151 gttttgcagc **aatgctggag cagaaaggat cacagatgtg gctttggatt**

Bold = EXON
Underline = sequence from for 5' splice site sequence and 3' splice site sequence, respectively from FIG. 9A

INTRON 4 DYT1

FIG. 17A: Clone 1:
Intron 4-5 prime.cDNA (Length: 535) SEQ ID NO.: 55

1 *GTCTGTGTCG GTTTTCAATA ACAAGAACAG* gTGAGTAGGG CCATCCACCG
51 CCAGTCCCAT CTGGTTCCTA ATCCTGCACC CTAAGTGTTA AAAGCATCAG
101 GGTCACTGTC AGCATCACCT GGGAGCTGGG TAGAAAGAAA TGgAGATTCT
151 CAGTCCCCTT CCGAGTCATG AGGGGAATCT TTGCTGATGA ACTCCAGGTA
201 ACTTTTATGA ACACTAATGT TTGACAAGTG CTGTTTTATT TTTATTTTT**C**
251 **AGATAGTTTT ACTCTGTCAC C**TAGGCTGGA GTGCAGTGGC GTAACCTTGC
301 CTCACTGCAA CCTCTGCCTC CCGGGCTCAA GCGATTCTTG TGCCTCAGCC
351 TCCTGAGTAG CTGGGATTAC AGGTGCACAC CATGCCCAAG CNAATATTTT
401 GTATTTTTAG TAGAGANGGG GCCCCGTNCA TGTTAACCAG GCTGGTCTTG
451 AACTNTTACC TCAGGTGAGT CCNCCACCTC GGCCTCCCAA AGTGCTGGGA
501 TTACAGGCGT GAGCCACTGT GTCTCAGCTT ATTTTT

Italics = EXON sequence
Underline = 5' splice sequence from FIG. 9A
Bold = primer 8 from FIG. 12A

FIG. 17B: Clone 2:
Intron 4-3 prime.cDNA (Length: 1302) SEQ ID NO.: 56

1 GCCACTCCAA GCTACCATCT GAGATTGTTT CCTGCCCTAG AGTGGTAAAG
51 GCGTGAGGTC CGTCTGCCCT CAGCTGTGTC CCCAGGCCCA GGGCGTGCCT
101 GGCAACANNA GCAGGCCTCT GAGAACCAGC CTCCCACGTG AGTTCATGAT
151 AGNAAGACAG CCCCTCGTTC CCATTCAGTG GTTGGTTCTG TTCTTTYCCT
201 GGCMATAAGC TCCACTCTGY MRTCAGCCAM ACATTTATTG AGTACCAGTT
251 GTTGGCAAAG CACTGTTGGG CATGAAAAGC ATTAACCCAG TGAATGAGGA
301 GGAGCTTGGG TTGGGACGGA GCCMCARAAW TACATGGCAG ACCAGAAGGA
351 AATCAGCTCA AGTAGAAARA CACGCATGGG CTCGTGGGCG ACGCAGTGTG
401 TGCTGTGTCA TCTGGGGCTG GGAGGAAGTG TCCTGGATCA GGAGTTCCAG
451 GAGCCCAGGA GGAGTGGACG GGTCAGTGCA GAGCCAGCCC GCAATCAGGG
501 GAAGAAAACA CGGCCAAGGC CAGGCCTTCA CGGGGAGCCC AGCGTGGGCT
551 GCACATCTGC ACTCTCCAGG CTAGTTTTGG TGCCCACATG CTCTGCAGGG
601 TCTGGGCACT GTGGCAGCGG CAGCAGGCTT CCCTGTTGCT AGTCCAGCTG
651 CTGAAACTCC AGGGAGAGTC AAAAAGTTCC CAAATACAGA GGCGTGGCTG
701 GTAGTCCTTC CCGGGAATTC TTCTTGCTTC CCGCTTTCTG TGGAACTCTG
751 CCTTCCCCAC TCTGCCTCTC TGCTTGTTCC TGGGCCCCAG GACCTCTTTC
801 CCATCTTCGA TCTCTTAAGT CATACCTTGG GAGGCCTCCC CCAGCCCGCC
851 GTGTAAAGAG GGCTGTCACA GCTTCTGCTG TCACAGAAGC ATTACAATGT
901 GCAGGTGCCT GTTAACATCT GCCTTCCCCA CTGATCTGGA GCTCCACAAG
951 GGAGAGGGCA CACCCAGTAG GTATGTGTGG GATGGATAGG AGGGTGGATG
1001 ACACCCAGTA GATGTGTATG GGATGGATAG GAGGGTGGAT GACACCCAGT
1051 AGGTGTGTAT GGGATGGATG GGAGGGTGGG TGACCCCTAG TAGATGTGGG
1101 GGGGGTGGGT GGGTGACCCC CAGTAGGTGT GTGTGGCATG GATAGGT**GAC**
1151 **CCCCAGTAGA CGTTTGT**GGG ACGGATGGGA GGGTAGGTAA GTGACCCCCA
1201 GGAGGCGTCT ATAGGGCAGG TGGGTGGATG TGGATGAACA GCACCTTGTT
1251 TCTTCTTCCC AG*GTGGCTTC TGGCACAGCA GCTTAATTGA CCGGAACCTC*
1301 AT

Bold = primer 9 in FIG. 12A
Underline is 3' splice site sequence from FIG. 9A
Italics is EXON 5 sequence

INTRON 1 5' from TORB

FIG 18A: Clone 1:
h59-29a5.se (Length: 240) SEQ ID NO.: 57

```
1    ggagcggccg ctcaacgctt cgggtacggc gcgcgcgcga gctgtgggtc
51   ggcgctgcgg ggggcgcggg ggcgcggggg cgcggaggga cggcctcgtg
101  ggcgcctggc acggaccggg cccgtggcat ctagacggcg gtggtcccag
151  ctggggtggg cggggagcgg atggggcggc cccggaaccg ttcgcnggaa
201  cgcagaagcn gtgccttgaa acactctcag atcgtgnggc
```

INTRON 1 3' from TORB

FIG. 18B: Clone 2:
5667s-29a5.Se (Length: 310) SEQ ID NO.: 58

```
1    gggaccaaag gacgtccgtc gttcccaccg accctaatcg ttcgcgngtc
51   ngttcgctac ccagtagaga gacttactta cnngtnnatc gaaggaatag
101  tctgggg[cttcgcaattcct ggaggtg]tat tagaactttc accgtagcaa
151  actgacggag ccgggatccc acaccgcctg tgggnncgac acgggaccta
201  ttgacacgaa gaacgaaacn gtcgattctt tcacgacgca acgactacgt
251  aaaaattcca gacaaagaga gaaacaagac cccgacaaga acgtcGAGAG
301  TTCGACCTAA
```

Upper Case Letters = EXON (bottom strand)
Underline = sequence from Table 1
Bold = 2nd primer from FIG. 12B

INTRON 2 5' from TORB

FIG. 19A: Clone 1:
6101-29a5.Se (Length: 401) SEQ ID NO.: 59

```
1    CAGGAACAAC AAAAATCCCA AGAAACCACT GACCCTTTCC TTACACGGCT
51   GGGCTGGCAC AGGCAAGAAT TTTGTCAGTN AAATTGTGGC TGAAAATCTT
101  CACCCAAAAG GTCTGAAGAG TAACTTTGTC CACCTGTTTG TATCGACTCT
151  GCACTTCCCT CATGAGCAGA AGATAAAACT GTACCAGgca agagaaccccg
201  ctattatctc gtctgcaggc cagtcggact ggtccgggtg acctgctcac
251  taactctggc ctctgcttct ctttcctttg tgttgctgta gcccccggct
301  ccactgagtt aaggcacact tagtccaggt agttacaaag ctctcctaca
351  acatttctta cttggttcca aaacagtcca gtggggtagg ggatgttatt
401  t
```

Upper Case Letters = EXON
Underline = 5'splice site sequence from FIG. 9B
Bold = 1st primer from FIG. 12B

INTRON 2 3' TORB

FIG. 19B: Clone 2:
29A5-39-11.se (Length: 238) SEQ ID NO.: 60

```
1    ttctgtaact ggtcCTGGAC CAACCATGAA AGAAGAAACA GGATGCGAAG
51   CTCAAAGGGC TGCACCAAGA GGCGCGCAGG CTCCATCTGC TCCTCATGCA
101  CTGAAGGACG AGGTCAGAGC TCTTAGAATG GCACCCTCAC CCCCACTCGC
151  TAGGTAGCAG CTTTTCTAAA ACCTTATCTC TAAAAAGTGG AAATTGGCAG
201  AGATAGATGC TAAAATGCAG AGAAGTTTTT CCTAACTC
```

Lower Case Letters = EXON
Underline = sequence from Table 1
Bold = primer 3 from FIG. 12B

INTRON 3 5' TORB

FIG. 20A: Clone 1:
39-14-29a5.Se (Length: 391) SEQ ID NO.: 61

```
1    GGGATCATTG ACGCAATCAA GCCGTTTCTA GACTACTACG AGCAGGTTGA
51   CGGAGTGTCT TACCGCAAAG CCATCTTCAT CTTTCTCAGg tcagcgggag
101  gcggtttttt ggggcacaca agcccttcat tctctcaatg ataaaatgag
151  gtcctgagga ccatcagcac tttgtttacc aggacgaaag tgcctgcttg
201  gcacaaggca cttacctact gctttacttt tcctttgcca gtcatcagca
251  tggcacacag tgtgggttgt ggaaatgaac taaagaaata atcactggga
301  caggcgcggt ggctcacacc tgtcaatccn agcactttgg gnaggcatgg
351  cgggcggatc acaggagatc gagacatctg ctaacatgnt g
```

Upper Case Letters = EXON
Underline = 5' splice site sequence from FIG. 9B
Bold = primer 4 from FIG. 12B

INTRON 3 3' TORB

FIG. 20B: Clone 2:
5665s-54a5.Se (Length: 373) SEQ ID NO.: 62

```
1    gtaagacaca gagtcttttt tnttttttag accgagtntc attnttgttg
51   ccnangctgg agtgcaatgg catgatctcg gctcgctgca acctccacct
101  cccggrttca aacgattctc ccacctcagc ctcccatgta gctgggatta
151  cagncatgca ccaccattag cctggctaat ttttgtgttt ttagtagaga
201  tggggttact ctatgttggt caggctggcc ttgaactccc gacctcaggt
251  gatctacctg cctcggcctc ccaaagtgct gggattacag ccatgagcna
301  ccacnscnan cagacncaga agtcttaata tgtgatttta atctttattt
351  ctctggcaaa ctcagCAATG CAG
```

Upper Case Letters = EXON
Underline = 3' splice site sequence from FIG. 9B
Bold = primer 5 from FIG. 12B

INTRON 4 TORB

FIG. 21: Clone 1:
intron4torb.se (Length: 310) SEQ ID NO.: 63

1 gtgagtccac cagggtaaag gagcccctta actgtccagc agtgagccgt
51 ctgctctttc attgagtgtt tgcacaaagc cacaggatcc cactggattt
101 cctcactttg ctaaagtcag gaattttctt agggcatact gtgctagaaa
151 ccagtgagtg agtgtccagc tgagtcctcg atgggcttgt tgcacactga
201 caagagacnc tctcaagggg tacggacatg aggaatgtgc tgagggtcgg
251 gactggagct tggccaggtg gcggtggtgg caggaaaccc agctgtgtct
301 tgttctgcag

Underline = 5' splice site sequence from FIG. 9B
Bold = primer 6 from FIG. 12B
Underline italics = 3' splice site sequence from FIG. 9B
Bold italics = primer 7 from FIG. 12B